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## Ophthalmologic Cystinosis

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*Introduction-* Cystinosis is a very rare lysosomal, autosomal recessive disease (1/200,000 births) caused by a mutation in the CTNS gene (Chz 17) encoding a protein called cystinosine. The intralysosomal accumulation of cystine induces the formation of insoluble crystals responsible for progressive multiple organ failure. Cystinous nephropathy is manifested by failure to thrive, Fanconi syndrome, damage to the renal glomerulus and manifestations affecting other organs appearing as early as 6 to 12 months of life. The specific treatment for cystinosis is cysteamine. The management is multidisciplinary.

We report the case of a boy, aged 6 years, followed for cystinosis diagnosed at the age of 18 months with polyuropolydipsic syndrome. The patient has been treated with oral cysteamine (Cystagon) since the age of 2 years.

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## INTRODUCTION

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We report the case of a boy, aged 6 years, followed for cystinosis diagnosed at the age of 18 months with polyuropolydipsic syndrome. The patient has been treated with oral cysteamine (Cystagon) since the age of 2 years.

On the ophthalmological level, his corrected visual acuity is evaluated at 10/10 P2 ODG, and the intraocular pressure is at 12 mmHg in both eyes. The main complaint of this patient is photophobia. He is treated with cysteamine eye drops (Cystadrops®) at a dosage of 4 drops per day and additional treatment with artificial tears. With the slit lamp, we find a corneal cystinosis of grade 1.50 according to the classification of Gahl (figure 1): deposits of birefringent spindle-shaped crystals accumulating in the corneal stroma, and progressing from the anterior stroma to the endothelium. The appearance is sparkling and multicolored on biomicroscopic examination.

The severity of the corneal involvement can be assessed by corneal OCT.

The fundus does not show retinopathy. Cystinosis retinopathy is very inconstant. Crystal deposits are sometimes found all over the retina, associated with depigmentation and which can progress to retinal atrophy. It can be explored by retinal angiography. The crystals can also deposit on the conjunctiva, iris, ciliary body, anterior lens capsule, choroid or optic nerve and cause various rarer manifestations, such as glaucoma by closing the angle, papillary edema or visual field changes.

Ophthalmologic monitoring for cystinosis should be annual, with visual acuity assessment, Gahl score estimate supplemented by OCT imaging of the cornea, measurement of intraocular pressure and fundus examination.

This case illustrates a well-followed patient who is coping well with his illness.

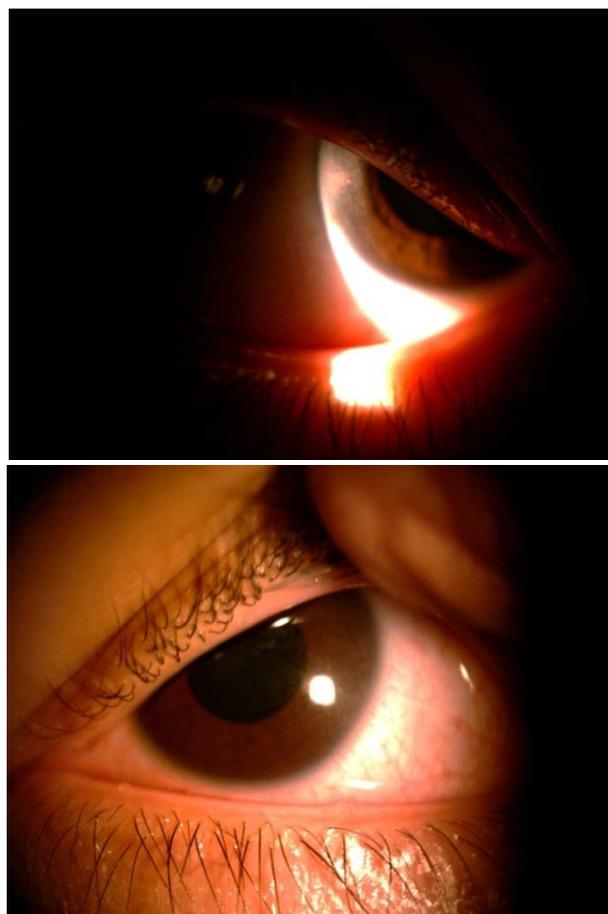


Figure 1: Corneal crystal deposits.

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